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ABSTRACT

Each year, state newborn screening programs test 4 million newborns for disorders that require early detection and treatment to prevent serious illness or death. The U.S. General Accounting Office (GAO) was asked to provide Congress with information on variations among state newborn screening programs. Based on surveys of such programs in all 50 states and the District of Columbia, the GAO found the following: (1) while the number of genetic and metabolic disorders included in state screening programs ranges from 4 to 36, most states screen for 8 or fewer disorders; (2) in deciding which disorders to include, states generally consider similar criteria, such as whether the disorder is treatable, and costs of screening for additional disorders; (3) most programs have similar practices for administering and funding their programs, and almost all provide education for parents and providers, but fewer than one-fourth inform parents of their option to obtain tests for additional disorders not included in the state's program; (4) state programs are primarily funded through fees collected from health care providers; (5) all newborn screening laboratories participate in a quality assurance program offered by Department of Health and Human Services' Centers for Disease Control and Prevention; (6) all states require newborn screening, and state statutes usually do not require parental consent, but 33 states allow exemptions from screening for religious reasons, and 13 additional states allow exemptions for any reason; and (7) newborn screening statutes and regulations in over half the states contain confidentiality provisions, but these provisions are often subject to exceptions. (Seven appendices include the scope and methodology of the GAO study, information on disorders most commonly included in state newborn screening programs, a chart of newborn screening program fees and expenditures per infant screened, and comments from the Department of Health and Human Services.) (HTH)

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GAO

Report to Congressional Requesters

March 2003

NEWBORN SCREENING

Characteristics of State Programs

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Highlights

Highlights of GAO-03-449, a report to Congressional Requesters

March 2003

NEWBORN SCREENING

Characteristics of State Programs

Why GAO Did This Study

Each year state newborn screening programs test 4 million newborns for disorders that require early detection and treatment to prevent serious illness or death. GAO was asked to provide the Congress with information on the variations among state newborn screening programs, including information on criteria considered in selecting disorders to include in state programs, education for parents and providers about newborn screening programs, and programs' expenditures and funding sources. To collect this information, GAO surveyed newborn screening programs for genetic and metabolic disorders in all 50 states and the District of Columbia. GAO was also asked to provide information on efforts by the Department of Health and Human Services (HHS) and states to evaluate the quality of newborn screening programs, state laws and regulations that address parental consent for newborn screening, and state laws and regulations that address confidentiality issues.

www.gao.gov/cgi-bin/getrpt?GAO-03-449.

To view the full report, including the scope and methodology, click on the link above. For more information, contact Marjorie Kanof at (202) 512-7119.

What GAO Found

While the number of genetic and metabolic disorders included in state newborn screening programs ranges from 4 to 36, most states screen for 8 or fewer disorders. In deciding which disorders to include, states generally consider similar criteria, such as whether the disorder is treatable. States also consider the cost of screening for additional disorders. HHS's Health Resources and Services Administration is funding an expert group to assist it in developing a recommended set of disorders for which all states should screen and criteria for selecting disorders.

Most state newborn screening programs have similar practices for administering and funding their programs. Almost all states provide education on their newborn screening program for parents and providers, but fewer than one-fourth inform parents of their option to obtain tests for additional disorders not included in the state's program. State programs are primarily funded through fees collected from health care providers, who may receive payments from Medicaid and other third-party payers. Nationwide, fees funded 64 percent of states' 2001 fiscal year program expenditures of over $120 million.

All newborn screening laboratories participate in a quality assurance program offered by HHS's Centers for Disease Control and Prevention, which assists programs in evaluating the quality of their laboratories. All states require newborn screening, and state statutes that govern screening usually do not require parental consent. However, 33 states' newborn screening statutes or regulations allow exemptions from screening for religious reasons, and 13 additional states' newborn screening statutes or regulations allow exemptions for any reason. Newborn screening statutes and regulations in over half the states contain confidentiality provisions, but these provisions are often subject to exceptions.

HHS said that the report presents a thorough summary of state newborn screening programs' current practices.

Disorders Most States Included in Their Newborn Screening Programs as of December 2002

Disorder	Number of states*
Phenylketonuria	51
Congenital hypothyroidism	51
Galactosemia	50
Sickle cell diseases	44
Congenital adrenal hyperplasia	32

Source: National Newborn Screening and Genetics Resource Center.

Note: This table does not include states that provide screening for the disorders to selected populations, as part of pilot programs, or by request.

*"States" refers to the 50 states and the District of Columbia.

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Abbreviations

AAP	American Academy of Pediatrics
APHL	Association of Public Health Laboratories
CDC	Centers for Disease Control and Prevention
CLIA	Clinical Laboratory Improvement Amendments of 1988
CMS	Centers for Medicare & Medicaid Services
CORN	Council of Regional Networks for Genetic Services
HHS	Department of Health and Human Services
HRSA	Health Resources and Services Administration
MCAD	medium-chain acyl-CoA dehydrogenase deficiency
MS/MS	tandem mass spectrometry
NCSL	National Conference of State Legislatures
NIH	National Institutes of Health
NSQAP	Newborn Screening Quality Assurance Program
PKU	phenylketonuria

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United States General Accounting Office
Washington, DC 20548

March 17, 2003

The Honorable Christopher J. Dodd
The Honorable Mike DeWine
United States Senate

Each year newborn screening programs in all the states test 4 million newborns to identify those who may have specific genetic and metabolic disorders that could threaten their life or long-term health.[1] Early detection, diagnosis, and treatment of these disorders may prevent a child's death, serious illness, or disability. For example, children with the metabolic disorder phenylketonuria (commonly referred to as PKU) cannot properly metabolize common foods, including milk and meat, and need to be placed on a special diet to avoid mental retardation. Children with sickle cell diseases, which are genetic blood disorders, can receive antibiotic treatment to reduce the risk of bacterial infections.

Newborn screening is a state public health activity, with each state responsible for designing and implementing its own program. For example, each state decides which disorders to include in its screening program. To assist the Congress as it considers actions related to newborn screening, you asked us to provide information on the variations among state newborn screening programs. In response to your request, this report provides information on (1) the disorders tested for in each state; how disorders are selected, including the use of advisory committees; and how states educate parents and health care providers about newborn screening, notify them of screening results, and follow up on abnormal results, (2) state newborn screening programs' expenditures and funding sources, (3) efforts by the Department of Health and Human Services (HHS) and states to monitor and evaluate the quality of state newborn screening programs, and (4) how state laws address consent and privacy issues related to newborn screening. As you requested, this report focuses only on newborn screening for genetic and metabolic disorders and does not include information on screening programs for hearing and infectious diseases.

[1]In this report, "states" refers to the 50 states and the District of Columbia.

To provide information on state newborn screening programs, we surveyed state health officers in all the states during October and November 2002. The survey collected information on the laboratory and program administration/follow-up components of states' newborn screening programs, including their expenditures and funding sources. For the purposes of the survey and this report, follow-up activities include activities that are provided in response to abnormal screening results, such as confirmation of diagnosis and referral for treatment. We did not ask for information on disease management and treatment services. We spoke with staff of several states' newborn screening programs to clarify survey responses and to obtain additional, more detailed information. We also reviewed information compiled by the National Newborn Screening and Genetics Resource Center, a project funded by HHS's Health Resources and Services Administration (HRSA), which collects information on state newborn screening programs. In addition, we reviewed documents and interviewed Centers for Disease Control and Prevention (CDC) and HRSA staff on their efforts to monitor and evaluate the quality of state newborn screening programs. To determine how state laws address consent and privacy issues related to newborn screening, we analyzed state statutes and selected regulations that provide for newborn screening for genetic and metabolic disorders, and state statutes that relate to privacy of genetic information generally. To identify state newborn screening statutes and regulations and state genetic privacy statutes, we relied on research material provided by the National Conference of State Legislatures. (For additional information on our scope and methodology, see app. I.)

We conducted our work from June 2002 through March 2003 in accordance with generally accepted government auditing standards.

Results in Brief

While the number of genetic and metabolic disorders included in state newborn screening programs ranges from 4 to 36, most states screen for 8 or fewer disorders. Authority for deciding which disorders to include in programs often rests with state health departments or boards of health, which generally receive input from advisory committees. Screening for certain disorders may also be mandated by state law. In deciding which disorders to include in their programs, states generally consider similar criteria, such as how often the disorder occurs in the population, whether an effective screening test exists, and whether the disorder is treatable. States also reported that they consider the cost of screening for additional disorders, which may include costs associated with performing more tests, acquiring and implementing new technology, and following up on

abnormal results. With the exception of federal recommendations that newborns be screened for PKU, congenital hypothyroidism, and sickle cell diseases, there are no federal guidelines on the set of disorders that should be included in state screening programs. HRSA is funding an expert group to assist it in developing a recommended set of disorders for which all states should screen and criteria for selecting disorders. Almost all states provide education on their screening program for parents and providers. However, fewer than one-fourth of the states inform parents of their option to obtain testing for additional genetic and metabolic disorders not included in the state's program. All state programs notify a health care provider, such as a physician or hospital, of abnormal newborn screening results; fewer than half routinely notify parents directly of abnormal results. All states also follow up on abnormal results; their follow-up activities may include obtaining additional laboratory information, referring the infant for treatment, or confirming that treatment has begun.

States spent over $120 million on newborn screening in their 2001 fiscal year, with most states spending from $20 to $40 for each infant screened. Most of these expenditures supported the laboratory component of screening programs, including the processing and analysis of specimens. Nationwide, newborn screening fees funded 64 percent of programs' expenditures. The fees are generally paid by the health care providers submitting specimens, who in turn may receive payments from Medicaid and other third-party payers, including private insurers. Other funding sources included HRSA's Maternal and Child Health Services Block Grant, direct payments from Medicaid, and other state and federal funds.

CDC and HRSA offer services to help states monitor and evaluate the quality of their newborn screening programs. All laboratories that perform testing for state newborn screening programs voluntarily participate in CDC's Newborn Screening Quality Assurance Program (NSQAP). This enables them to meet the federal regulatory requirement under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to have a process for verifying the accuracy of tests they perform. HRSA's National Newborn Screening and Genetics Resource Center conducts technical reviews of individual state newborn screening programs that request them; the Resource Center has conducted nine reviews since January 2000. These reviews respond to specific questions raised by state officials, such as how to implement an expansion of the state's program. The reviewing team also analyzes the overall state newborn screening program and provides the state with findings and recommendations that could improve the program. States are not obligated to implement these recommendations. In addition to participating in federal quality assurance programs, most state

newborn screening programs reported that they evaluate the quality of the laboratory testing or program administration/follow-up components of their programs.

State newborn screening statutes usually do not require that parental consent be obtained before screening occurs. While all states require newborn screening, 33 states' newborn screening statutes or regulations allow exemptions from screening for religious reasons, and 13 additional states' newborn screening statutes or regulations allow exemptions for any reason. Newborn screening statutes and regulations in over half the states specify that newborn screening information is confidential, but these confidentiality provisions are often subject to exceptions, which vary across states. The most common exception allows disclosure of information for research purposes, provided that the child's identity is not revealed and researchers comply with applicable laws for the protection of humans in research activities. Other exceptions include use of information for law enforcement and for establishing paternity. Over half the states have statutes that govern the collection, use, or disclosure of genetic information, which may also apply to genetic information obtained from newborn screening. While few newborn screening statutes provide penalties for violation of confidentiality provisions, 17 states' genetic privacy statutes provide specific penalties for violating genetic privacy laws.

In commenting on a draft of this report, HHS said that the report presents a thorough summary of state newborn screening programs' current practices.

Background

Newborn screening programs in the United States began in the early 1960s with the development of a screening test for PKU and a system for collecting and transporting blood specimens on filter paper. All newborn screening begins with a health care provider collecting a blood specimen during a newborn's first few days of life.[2] The baby's heel is pricked to obtain a few drops of blood, which are placed on a specimen collection card and sent to a laboratory for analysis. State departments of health may

[2]All states have screening statutes or regulations that specify certain health care providers who are responsible for ensuring that newborns are screened, such as the attending physician, nurse, midwife, hospital, or other institution caring for the infant. Some state screening statutes and regulations include a child's parent among those who are responsible for ensuring that screening occurs.

use their own laboratory to test samples from the dried blood spots or may have a contract with a private laboratory, a laboratory at a university medical school, or another state's public laboratory.

Laboratories may choose among a variety of testing methods to maximize the efficiency and effectiveness of their testing. A major technical advance in newborn screening is use of the tandem mass spectrometer, an analytical instrument that can precisely measure small amounts of material and enable detection of multiple disorders from a single analysis of a blood sample. Tandem mass spectrometry (MS/MS) has greatly increased the number of disorders that can be detected, but it cannot completely replace other analysis methods because it cannot screen for all disorders included in state newborn screening programs.

After initial testing, state newborn screening program staff notify health care providers of abnormal results because it may be necessary to verify the accuracy of the initial screening result by testing a sample from a second specimen or to ensure that the infant receives more extensive diagnostic testing to confirm the presence of a disorder. The infant may also need immediate treatment. Laboratories and state maternal and child health programs generally carry out the notification process.

Primary care and specialty physicians are involved in various stages of the newborn screening process. They generally are responsible for notifying the family of abnormal screening results and may confirm initial results through additional testing. If necessary, they identify appropriate management and treatment options for the child. State maternal and child health program staff may follow up to ensure that these activities occur.

Federal Role in Newborn Screening

Several HHS agencies carry out activities related to newborn screening, including collecting and sharing information about state newborn screening programs, promoting quality assurance, and funding screening services. HRSA's Maternal and Child Health Bureau has primary responsibility for promoting and improving the health of infants and mothers. HRSA offers grants to states, including the Maternal and Child Health Services Block Grant, that state newborn screening programs may use to support their newborn screening services. HRSA also funded the development of the Council of Regional Networks for Genetic Services (CORN) in 1985 to provide a forum for information exchange among groups concerned with public health aspects of genetic services. The newborn screening committee of CORN identified several areas of importance to programs, including the process of selecting disorders for

screening, communication, quality assurance, and funding. It developed guidelines in these areas to increase consistency among state newborn screening programs[3] and also began collecting data on state programs. In 1999, CORN was disbanded, and HRSA established the National Newborn Screening and Genetics Resource Center—the Resource Center. The Resource Center is supported by a cooperative agreement between the Genetic Services Branch of HRSA's Maternal and Child Health Bureau and the University of Texas Health Science Center at San Antonio Department of Pediatrics. The Resource Center develops annual reports on state newborn screening activities and provides technical assistance to state newborn screening programs. It also provides information and educational resources to health professionals, consumers, and the public health community.

CDC's Newborn Screening Branch,[4] in partnership with the Association of Public Health Laboratories (APHL), operates NSQAP.[5] NSQAP is a voluntary, nonregulatory program that is designed to help state health departments and their laboratories maintain and enhance the quality of their newborn screening test results. In addition, CDC's National Center on Birth Defects and Developmental Disabilities funds research related to newborn screening.

The Centers for Medicare & Medicaid Services' (CMS) involvement in newborn screening relates to its Medicaid and CLIA programs. CMS administers Medicaid, a jointly funded, federal-state health insurance program for certain low-income individuals, which covers newborn screening for eligible infants. Nationwide, Medicaid finances services for one in three births each year. Through the CLIA program,[6] CMS also

[3]Council of Regional Networks for Genetic Services, "U.S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetic Services," *Screening*, vol. 1 (1992). Additional CORN guidelines were published in 2000; see Council of Regional Networks for Genetic Services, "U.S. Newborn Screening System Guidelines II: Follow-up of Children, Diagnosis, Management, and Evaluation—Statement of the Council of Regional Networks for Genetic Services," Supplement to *The Journal of Pediatrics*, vol. 137, no. 4 (2000).

[4]The Newborn Screening Branch is in the National Center for Environmental Health's Department of Laboratory Services.

[5]NSQAP has a memorandum of understanding with APHL. APHL provides assistance to NSQAP on how the program operates, including input on how to report data and which disorders to include in NSQAP.

[6]Pub. L. No. 100-578 § 2, 102 Stat. 2903, 2907.

regulates laboratory testing performed on specimens obtained from humans, including the dried blood spots used for newborn screening. CLIA's purpose is to ensure the accuracy, reliability, and timeliness of laboratory test results. CLIA requires that laboratories comply with quality requirements in five major areas: personnel qualifications and responsibilities, quality control, patient test management, quality assurance, and proficiency testing.[7] Laboratories that fail to meet CLIA's quality requirements are subject to sanctions, including denial of Medicaid payments.[8] Through the CLIA program, laboratories that test dried blood spots in connection with newborn screening must have a process for verifying the accuracy of their tests at least two times each year. State newborn screening laboratories can meet this requirement through participation in the proficiency testing program offered by NSQAP.

The National Institutes of Health's (NIH) National Institute of Child Health and Human Development has sponsored research on disorders identified through newborn screening, including PKU, congenital hypothyroidism, and galactosemia. Research has addressed issues such as the effectiveness of screening and treatments and the application of new technologies for identifying additional disorders.

The Children's Health Act of 2000 authorized HHS to award grants to improve or expand the ability of states and localities to provide screening, counseling, or health care services for newborns and children who have, or are at risk for, heritable disorders and to evaluate the effectiveness of these services.[9] As of February 2003, funds had not been appropriated to fund these grants. The act also authorized the establishment of a committee to advise the Secretary of HHS on reducing the mortality and morbidity of newborns born with disorders. The Secretary of HHS signed the charter for this committee in February 2003.

Federal Privacy Standards

Under the Health Insurance Portability and Accountability Act of 1996,[10] HHS developed regulations to protect the privacy of health information,

[7]Proficiency testing is the process of sending sample specimens to laboratories to verify the accuracy and reliability of their tests.

[8]This could also result in denial of Medicare payments.

[9]Pub. L. No. 106-310, § 2601, 114 Stat. 1101, 1164.

[10]Pub. L. No. 104-191 § 264, 110 Stat. 1939, 2033-2034.

which as defined in the regulations, would include the results of testing of newborns. The regulations give individuals the right, in most cases, to inspect and obtain copies of health information about themselves. In addition, the regulations generally restrict health plans and certain health care providers from disclosing such information to others without the patient's consent, except for purposes of treatment, payment, or healthcare operations.[11] While the federal regulations preempt state requirements that conflict with them, states are free to enact and enforce more stringent privacy protections. Most entities and individuals that are covered by the regulations must be in compliance by April 14, 2003.

Disorders Included in State Newborn Screening Programs Vary, but Administration of Program Components Is Similar

Although state newborn screening programs vary in the number of disorders for which they screen, states generally follow similar practices and criteria in selecting disorders for their programs. States also conduct most other aspects of their programs in similar ways. Almost all state programs provide information for parents and conduct provider education, but fewer than one-fourth of the states provide information for parents on their option to test for additional disorders not included in the state's program. All state programs notify health care providers—and some also notify parents—about abnormal screening results, and all states reported following up on abnormal results.

Most States Screen for Eight Disorders or Fewer

Most state newborn screening programs screen for 8 disorders or fewer. The number of disorders included in state programs ranges from 4 to 36. (See app. II for the number of disorders screened for by each state.) Programs are implemented through state statutes and/or regulations, which often require screening for certain disorders. According to the Resource Center, all states require screening for PKU and congenital hypothyroidism, and 50 states require screening for galactosemia. Table 1 lists the disorders most commonly included in state newborn screening programs. (See app. III for information on these disorders.) Some states provide screening for certain disorders to selected populations, through pilot programs, or by request. For example, in addition to the 44 states that require screening for sickle cell diseases for all newborns, 6 states provide screening for sickle cell diseases to selected populations or through pilot

[11]There are additional exceptions to facilitate compliance with state reporting requirements and other public health purposes.

programs. Some states are taking steps that could expand the number of disorders included in their programs.[12]

Table 1: Disorders Most Commonly Included in State Newborn Screening Programs, December 2002

Disorder	Number of states*
PKU	51
Congenital hypothyroidism	51
Galactosemia	50
Sickle cell diseases	44
Congenital adrenal hyperplasia	32
Biotinidase deficiency	24
Maple syrup urine disease	24
Homocystinuria	17

Source: National Newborn Screening and Genetics Resource Center.

Note: This table does not include states that provide screening for the disorders to selected populations, as part of pilot programs, or by request.

*"States" refers to the 50 states and the District of Columbia.

The criteria that state newborn screening programs reported they consider in selecting disorders to include in their programs are generally consistent across states. For example, they generally include how often the disorder occurs in the population, whether an effective screening test exists to identify the disorder, and whether the disorder is treatable. These criteria are also consistent with recommendations of the American Academy of Pediatrics (AAP) newborn screening task force.[13] Neither the criteria states use nor AAP's recommendations include benchmarks, such as the lowest

[12]For example, Connecticut, which screens for 8 disorders, plans to add 3 disorders to its program in March 2003 and is considering adding others. Mississippi, which screens for 5 disorders, is in the process of reviewing proposals from laboratories to conduct screening for 35 additional disorders. Virginia, which screens for 8 disorders, has added medium-chain acyl-CoA dehydrogenase deficiency (MCAD) to the state's newborn screening program, contingent on the program's acquiring funding to support follow-up staff and the purchase of necessary equipment. Children with MCAD cannot convert fat to energy, and must avoid fasting, which might occur when the child is ill. To avoid risk of seizures, brain damage, or death, these children must either continue eating while ill or receive nutrients under medical supervision.

[13]American Academy of Pediatrics Newborn Screening Task Force, "Serving the Family From Birth to the Medical Home: Newborn Screening: A Blueprint for the Future—A Call for a National Agenda on State Newborn Screening Programs," *Pediatrics*, vol. 106, no. 2 (2000). HRSA funded the task force.

incidence or prevalence rate that would be acceptable for population-based newborn screening or measurements of treatment effectiveness or screening reliability.

Some states reported that they are considering revising their criteria because MS/MS can identify disorders for which treatment is not currently available. Because MS/MS technology can be used for screening multiple disorders in a single analysis, states may choose to include such disorders in their testing along with disorders that can be treated.[14] Twenty-one states use MS/MS in their screening programs (see app. II);[15] the number of disorders for which screening is conducted using MS/MS ranges from 1 to 28. (See app. IV for a list of selected disorders for which screening is conducted using MS/MS.)

Many states consider cost when selecting disorders to include in their newborn screening program. In addition, several states told us that they would need additional funding to expand the number of disorders in their program. The costs associated with adding disorders include costs of additional testing, educating parents and providers, and following up on abnormal results. Additional costs may also be associated with acquiring and implementing new technology, such as purchasing MS/MS technology and training staff in its use.

With the exception of federal recommendations that newborns be screened for three specific disorders, there are no federal guidelines on the set of disorders that should be included in state screening programs. The U.S. Preventive Services Task Force, which is supported by HHS's Agency for Healthcare Research and Quality, has recommended screening for sickle cell diseases, PKU, and congenital hypothyroidism. In addition, NIH issued a consensus statement recommending that all newborns be screened for sickle cell diseases, as well as a consensus statement concluding that genetic testing for PKU has been very successful in the prevention of severe mental retardation.[16] AAP's newborn screening task

[14]There has been discussion among experts about the appropriate use of MS/MS in newborn screening. This has focused on several issues, including whether the incidence and severity of the disorders detected by MS/MS justifies screening and whether effective treatment would be available for disorders detected.

[15]Twelve additional states reported they plan to begin using MS/MS by the end of 2003.

[16]NIH consensus statements are prepared by a nonfederal panel of experts and reflect the panel's assessment of medical knowledge available at the time the statement is written.

force reported that infants born anywhere in the U.S. should have access to screening tests and procedures that meet accepted national standards and guidelines. The task force recommended that federal and state public health agencies, in partnership with health professionals and consumers, develop and disseminate model state regulations to guide implementation of state newborn screening systems, including the development of criteria for selecting disorders. In 2001, HRSA awarded a contract to the American College of Medical Genetics to convene an expert group to assist it in developing a recommended set of disorders for which all states should screen and criteria that states should consider when adding to or revising the disorders in their newborn screening programs.[17] The expert group is expected to make recommendations to HRSA in spring 2004. Some state officials told us they have concerns about the development of a uniform set of disorders because states differ in incidence rates for disorders and capacity for providing follow-up and treatment.

Most states reported that the state health department or board of health has authority to select the disorders included in newborn screening programs. Six states reported that they could not modify the disorders included in their newborn screening programs without legislation. Forty-five states reported that they have an advisory committee that is involved in selecting disorders; such a committee generally makes recommendations to the state health department or board of health. Most states reported that their advisory committee is not required by state statute or regulation. We found that most newborn screening advisory committees are multidisciplinary and include physicians, other health workers, and individuals with disorders or parents of children with disorders. (See table 2.)

[17]The expert group is also charged with recommending minimum standards for state newborn screening programs to use in assessing and evaluating their programs, and with recommending health outcomes that would be appropriate to use in monitoring and evaluating newborn screening. In addition, it is to consider the value of establishing a national process for the evaluation and oversight of newborn screening programs.

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Table 2: Categories of Individuals Represented on States' Newborn Screening Advisory Committees

Category	Number of states[a]
Specialty medical care physicians[b]	44
Laboratory specialists	41
Pediatricians and/or other primary health care providers	40
Health department staff who conduct follow-up activities	38
Individuals with disorders or parents of children with disorders	35
Ethicists	16
Other[c]	28

Source: GAO Survey of State Newborn Screening Programs for Genetic and Metabolic Disorders, October 21, 2002.

[a]Forty-four states and the District of Columbia reported that they have an advisory committee.

[b]Includes metabolic specialists, endocrinologists, geneticists, and hematologists.

[c]Includes representatives from state hospital associations, state March of Dimes chapters, social workers, lawyers, other state and local health department staff, dieticians, and state legislators.

Most States Provide Information for Parents and Conduct Provider Education, but Few Provide Information to Parents on Screening Not Included in State Program

Almost all states reported they offer information for parents and education for providers on their newborn screening program. Eleven states have newborn screening statutes requiring that parents of newborns be informed of the program at the time of screening.[18] In most states, information for parents includes how the blood specimen is obtained, the disorders included in the state program, and how parents will be notified of testing results. Seven states reported they include information for parents on their option to obtain testing for additional disorders that are not included in the state's program, but that may be available to them through other laboratories.[19] Provider education offered by states includes information on the collection and submission of specimens, the management of the disorders, and medical specialists available to treat the disorders.

[18]The 11 states are California, the District of Columbia, Delaware, Maryland, Missouri, Nebraska, New Mexico, Oregon, Vermont, Wisconsin, and Wyoming. Some of these state statutes require that specific information be provided to parents, such as the purpose of the screening and the risks involved. Other statutes do not specify the type of information that should be communicated to parents.

[19]Five of these states and five additional states reported that they communicate information to health care providers on parents' option to obtain testing for additional disorders that are not included in the state's program.

While state newborn screening programs produce or compile materials for parents, they generally do not provide them directly to parents and are unable to say when, or if, parents actually receive them. Rather, the state provides materials to other individuals, including hospital staff, midwives, pediatricians, primary care providers, and local health department staff, who are expected to share them with parents. Over half the states reported that their materials for parents are available in English and one or more other languages.

States Generally Notify Multiple Parties of Abnormal and Normal Screening Results and Follow Up on Abnormal Results

The parties states notify about newborn screening results vary, depending on whether the result is abnormal[20] or normal. (See table 3.) All states reported that for abnormal results, they notify the physician of record or the birth or submitting hospital. The physician or hospital, in turn, is generally responsible for notifying parents. Most states reported they notify physicians and hospitals by telephone; many states reported also notifying them by letter, fax, or E-mail. While the AAP newborn screening task force recommended that programs notify parents or guardians, fewer than half the states routinely notify parents directly of abnormal results, and no state routinely notifies parents directly of normal results. States that notify parents generally said that notification of parents was by letter.

Table 3: Number of States Notifying Specific Parties of Newborn Screening Results

	Number of states	
Party notified	**Abnormal results**	**Normal results**
Birth or submitting hospital	50	49
Physician of record	51	34
Specialty provider	34	[b]
Parent	22	0
Other[c]	16	7

Source: GAO Survey of State Newborn Screening Programs for Genetic and Metabolic Disorders, October 21, 2002.

[a]"States" refers to the 50 states and the District of Columbia.

[b]Because specialty care is not necessary for children with normal results, we did not ask states if a specialty provider was notified.

[c]Includes midwives, county and local health departments, and the infant's primary care physician.

[20]There are two types of abnormal results. Those that are strongly positive require the newborn to immediately receive diagnostic tests or treatment. Those for which the reliability of the result is questionable require testing of a sample from a second specimen, which is less time-critical.

States also reported that they take other actions in response to abnormal screening results. About three-fourths of states reported testing samples from second specimens when the initial specimen is abnormal or unsatisfactory.[21] All states reported conducting follow-up activities. Over 90 percent of states said that their follow-up activities include obtaining additional laboratory information to confirm the presence of a disorder, which could include obtaining the results of diagnostic tests performed by other laboratories. Almost all states reported that they refer infants with disorders for treatment and most follow up to confirm that treatment has begun. About two-thirds of the states reported that they conduct or fund periodic follow-up of newborns diagnosed with a disorder, which could include ensuring that they continue to receive treatment and monitoring their health status. According to Resource Center data on state newborn screening programs, the length of the follow-up period varies among disorders and across states.[22]

State Spending on Newborn Screening Varies, and Majority of State Programs Receive Most Funding from Fees

States reported that they spent over $120 million on newborn screening in state fiscal year 2001, with individual states' expenditures ranging from $87,000 to about $27 million. Seventy-four percent of these expenditures supported laboratory activities. The primary funding source for most states' newborn screening expenditures was newborn screening fees. The fees are generally paid by health care providers submitting specimens; they in turn may receive payments from Medicaid and other third-party payers, including private insurers. Other funding sources that states identified included the Maternal and Child Health Services Block Grant, direct payments from Medicaid, and other state and federal funds.

[21]One state reported that testing samples from second specimens is required if the first specimen is collected before the newborn is 48 hours old, regardless of whether the initial test result was normal or abnormal. Thirteen states reported testing samples from second specimens for all newborns for all tests included in the initial screen.

[22]National Newborn Screening and Genetics Resource Center, *National Newborn Screening Report - 1999*, (Austin, Tex.: July 2002).

Newborn Screening Expenditures Vary by State

States reported they spent over $120 million on laboratory and program administration/follow-up activities in state fiscal year 2001.[23,24] Individual states' expenditures ranged from $87,000 to about $27 million. Based on information provided by 46 states, we found that, on average, states spent $29.44 for each infant screened in state fiscal year 2001.[25] Two-thirds of these states spent from $20 to $40 per infant. (See app. V for expenditures per infant screened in each state.)

Laboratory expenditures accounted for 74 percent of states' expenditures; program administration/follow-up expenditures accounted for 26 percent.[26] States reported that laboratory expenditures generally supported activities such as processing and analyzing specimens, notifying health care providers and parents of screening test results, and evaluating the quality of laboratory activities. Program administration/follow-up expenditures generally supported activities such as notifying appropriate parties of test results, confirming that infants received additional laboratory testing, confirming that infants diagnosed with disorders received treatment, and providing education to parents and health care providers. In addition, almost half the states reported that laboratory expenditures supported education of parents and health care providers.

[23]We asked states to provide us expenditure information for laboratory and program administration/follow-up; we instructed states to include only those follow-up activities that are conducted through confirmation of diagnosis and referral for treatment. We did not ask for expenditure information for disease management and treatment services.

[24]Expenditure calculations were based on responses from 50 states; South Dakota reported that expenditure information was not available for state fiscal year 2001. Six states reported that their expenditures included significant, nonrecurring expenses in state fiscal year 2001, such as for the purchase of MS/MS equipment or computer software. These expenditures ranged from $22,645 to $415,835, totaling about $1 million. In addition, one state told us that the program administration/follow-up expenditures it reported included approximately $50,000 to $75,000 for disease management and treatment services.

[25]We were unable to calculate expenditures per infant screened for five states. South Dakota reported that expenditure information was not available for state fiscal year 2001. Florida, Georgia, Kentucky, and Minnesota did not provide information on the number of infants screened.

[26]Expenditure calculations are based on responses from 49 states. South Dakota reported that expenditure information was not available for state fiscal year 2001. New York provided total expenditure information but did not separately identify expenditures for the laboratory and program administration/follow-up components.

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State Newborn Screening Programs Are Funded Primarily through Fees

Fees are the largest funding source for most states' newborn screening programs. Forty-three states reported they charge a newborn screening fee to support all or part of program expenditures.[27] The fees are generally paid by health care providers submitting specimens; they in turn may receive payments from Medicaid and other third-party payers, including private insurers. Some states collect the fees through the sale of specimen collection kits to hospitals and birthing centers. Other states may bill hospitals, patients, physicians, Medicaid, or other third-party payers for the fee. Nationwide, newborn screening fees funded 64 percent of newborn screening program expenditures in state fiscal year 2001.[28,29] (See table 4.) Thirteen state programs reported that fees were their sole source of funding in fiscal year 2001, and 19 additional states reported that fees funded at least 60 percent of their newborn screening expenditures. As of The average fee in the states that charged a fee was about $31, with fees ranging from $10 to $60.

Table 4: Funding Sources for State Newborn Screening Programs, as Percentage of Nationwide Program Expenditures, State Fiscal Year 2001

Funding source	Percentage of program expenditures
Fees	64
Maternal and Child Health Services Block Grant	5
Medicaid[a]	10
Other state funds	19
Other funds[b]	2

Source: GAO Survey of State Newborn Screening Programs for Genetic and Metabolic Disorders, October 21, 2002.

Note: This table includes information for 50 states; South Dakota reported that information on state fiscal year 2001 funding sources was not available. We asked states to provide us expenditure information for laboratory and program administration/follow-up components and instructed them to include only those follow-up activities that are conducted through confirmation of diagnosis and referral for treatment. We did not ask for expenditure information for disease management and treatment services.

[a]Includes federal and state contributions.

[b]Includes, for example, the Preventive Health and Health Services Block Grant.

[27]We asked states to report whether they currently charge a fee, and if so, the amount of that fee. States responded to the survey in October and November 2002.

[28]States may have also used fees to support disease management and treatment activities.

[29]South Dakota is not included in any of the calculations related to funding sources; it reported that information on state fiscal year 2001 funding sources was not available.

Seven state newborn screening programs identified Medicaid as a direct funding source in state fiscal year 2001. These screening programs bill the state Medicaid agency directly for laboratory services or receive a transfer of funds from the state Medicaid agency for screening services provided to Medicaid-enrolled infants. The percentage of expenditures the states reported as directly funded by Medicaid does not include Medicaid payments to hospitals for services provided to newborns.[30]

Other funding sources that states identified for newborn screening program expenditures include state funds and the Maternal and Child Health Services Block Grant. About half the states reported that state funds supported laboratory or program administration/follow-up expenditures. In addition, about half the states reported that they rely on the Maternal and Child Health Services Block Grant as a funding source for laboratory or program administration/follow-up expenditures. Seven states identified other funding sources, such as the Preventive Health and Health Services Block Grant.

Newborn Screening Quality Assurance Efforts Focus on Laboratory Testing and Performance Monitoring

CDC and HRSA offer services to assist states in evaluating the quality of their newborn screening programs. For example, CDC's NSQAP provides proficiency testing for almost all disorders included in state newborn screening programs, enabling states to meet the CLIA regulatory requirement that laboratories have a process for verifying the accuracy of tests they perform. Through the Resource Center, HRSA supports technical reviews of state newborn screening programs. These voluntary programwide reviews are conducted at the request of state health officials and focus primarily on areas of concern identified by state officials. In addition to these federally supported efforts, most state newborn screening programs reported that they evaluate the quality of the laboratory testing and/or program administration/follow-up components of their newborn screening programs.

[30]Medicaid may reimburse hospitals for newborn screening services on a fee-for-service basis or as part of a maternity care package.

CDC Provides Proficiency Testing and Other Quality Assurance Services to Newborn Screening Laboratories

CDC's NSQAP is the only program in the country that conducts proficiency testing on the dried blood spots used in newborn screening.[31] While NSQAP is voluntary, as of January 2003, all laboratories that perform testing for state newborn screening programs participated in the proficiency testing program. Participation in NSQAP allows laboratories to meet the CLIA regulatory requirement that they have a process for verifying the accuracy of tests they perform. NSQAP offers proficiency testing for over 30 disorders, including the disorders most commonly included in state newborn screening programs.

When a laboratory misclassifies a specimen during proficiency testing, NSQAP notifies the laboratory of the problem. When an abnormal specimen is classified as normal, NSQAP officials work with the laboratory to identify and solve the problem that led to the misclassification. NSQAP provides information on the specimen that was misclassified, gives supplemental specimens to the laboratory to test, and may visit the laboratory, if necessary, to provide additional assistance.[32]

In addition to proficiency testing, NSQAP provides other types of quality assurance assistance, including training, guidelines, and consultation to laboratories that participate in the program. For example, in September 2001, NSQAP cosponsored a meeting of laboratory and medical scientists to discuss issues related to the use of MS/MS in newborn screening.[33] In addition, NSQAP provides state newborn screening programs with quality control specimens—test specimens designed to be run over a period of time to ensure the stability of the testing methods—and works with the manufacturers of the filter papers used in the collection of dried blood

[31]To conduct proficiency testing, NSQAP prepares and distributes specimens quarterly to participating laboratories. NSQAP does not include information on the expected results with these specimens. Laboratories analyze samples from the specimens and return their analytical results and clinical assessments to NSQAP for review. NSQAP compares the laboratory's results to the expected results for the specimen. All laboratories receive at least two abnormal specimens for each disorder for which they test during the course of the year. These proficiency testing services are provided at no charge to laboratories.

[32]According to NSQAP officials, when a laboratory misclassifies a normal specimen as abnormal, they inform the laboratory of the misclassification, but do not offer additional assistance. This misclassification is not considered a serious problem because the additional laboratory testing that should follow an abnormal screening result would confirm that the newborn does not have the disorder.

[33]NSQAP cosponsored this meeting with HRSA, APHL, and the Wisconsin Department of Health.

spots to ensure their quality.[34] NSQAP also publishes quarterly and annual reports on the aggregate performance of participating laboratories. These reports include information on the results of the proficiency testing program. The annual reports also include information on NSQAP's quality control effort and describe other activities undertaken during the year.

HRSA Funds Voluntary Technical Reviews of State Newborn Screening Programs

HRSA's Resource Center offers technical reviews to states at their request to help them refine and improve their newborn screening activities.[35] The team that visits the state program typically includes a representative of the Resource Center, a representative from CDC's NSQAP to focus on laboratory quality assurance, a health care provider to focus on medical and genetic issues, a follow-up coordinator from another state program to focus on the follow-up component of the program, and a representative from HRSA to focus on financial and administrative issues. The Resource Center's reviews concentrate primarily on areas state officials ask the team to review. For example, states have asked the review team to look at whether or how the set of disorders included in their programs should be expanded, how to incorporate MS/MS into a program, and whether current program staffing levels are appropriate. The review team also assesses the degree to which the state program follows the 1992 CORN guidelines in areas such as public, professional, and patient education, laboratory proficiency testing, and consumer representation on advisory committees.

After reviewing a state newborn screening program, the team provides the state with a final report that includes its findings and recommendations to improve the program. Recent findings have included newborn screening advisory committees that were not sufficiently multidisciplinary and programs that did not have a systemwide quality assurance program. Review teams have also identified the need for additional program administration/follow-up staff and for provider education programs to include information on collecting and submitting specimens and reporting screening results. The state newborn screening program is not obligated to accept or implement the team's recommendations, and HRSA and the Resource Center have no authority to require states to make changes to their program. However, according to the Resource Center, most participating states have made some modifications to their program in

[34]The manufacturers of the filter paper voluntarily send statistically valid sample sets of production lots for evaluation against specific NSQAP criteria.

[35]Prior to 1999, HRSA contracted with an expert panel to conduct these reviews.

response to recommendations. State officials told us, for example, that they have expanded or diversified the membership of their advisory committees, revised practitioner manuals, developed a programwide quality assurance system, and hired additional program administration/follow-up staff. In addition, state newborn screening program staff told us that the recommendations of the review teams helped inform program staff, state legislators, and health department staff as they assessed program needs.

HRSA has funded 26 technical reviews in 22 states since the program began in 1987;[36] 9 of these reviews have occurred since January 2000. Every state that has requested a review has been able to receive one.

Most State Newborn Screening Programs Reported Evaluating Laboratory or Program Administration/Follow-up Activities

Most states reported evaluating the quality of the laboratory testing and/or program administration/follow-up components of their newborn screening programs. For example, laboratories monitor performance by defining criteria for achieving quality results and designing a monitoring program to evaluate whether they are meeting these criteria. One state told us that it has criteria related to calibration of equipment, personnel training and education, and recordkeeping and documentation. Other measures that programs may monitor include percentage of births screened, number of unusable specimens, demographic information missing from specimen collection cards, and number of children lost to follow-up. Several state officials told us that they use some of these measures to monitor quality of specimens received from hospitals and to identify hospitals that may need education regarding the newborn screening process. In addition, states voluntarily report many of these measures to the Resource Center for inclusion in its annual National Newborn Screening Report, enabling states to compare their program over time with other states' programs. Moreover, all states report annually to HRSA on the percentage of newborns in the state who are screened for selected disorders, including PKU and congenital hypothyroidism, as part of the Maternal and Child Health Services Block Grant reporting requirements.[37]

[36]Four states requested a second review several years after receiving the first review. In addition to these 22 states, Guam and Saipan have also participated in the program.

[37]In addition, some states have developed other performance measures related to newborn screening, which they submit to HRSA as part of their Maternal and Child Health Services Block Grant annual report. For example, one state reports on the percentage of newborns with abnormal screening results who receive follow-up.

About half the states reported to us that they have a mechanism for learning of abnormal cases that were misclassified as normal, information that can alert a state to problems with its program. According to experts in the field of newborn screening, these cases occur infrequently but can have serious results when children develop a life-threatening condition that might have been prevented if treated early. Most of these states learn about these cases through their communications with the specialists in their state who manage and treat the disorders identified by newborn screening. If a child is referred to one of these specialists from a source other than the newborn screening program, the specialist will usually contact program officials, who then determine whether the screening program misclassified the child's screening result as normal. Four states reported that they can learn of abnormal cases misclassified as normal through reports made to state birth defects or disease registries. For example, one state reported that staff at the state birth defects registry notify the newborn screening program of children reported to them, and the newborn screening program then checks whether or not these children were identified through the screening process.

States Generally Do Not Require Consent for Newborn Screening and Most Limit Disclosure of Screening Information

State newborn screening statutes usually do not require that parental consent be obtained before screening occurs. However, most state newborn screening statutes or regulations allow exemptions from screening for religious reasons, and several states allow exemptions for any reason. Provisions regarding the confidentiality of screening results are included in state newborn screening statutes and regulations and state genetic privacy laws, but are often subject to exceptions, which vary across states. The most common exceptions allow disclosure of information for research purposes, for use in law enforcement, and for establishing paternity. While few newborn screening statutes provide penalties for violation of confidentiality provisions, many states' genetic privacy statutes provide criminal sanctions and penalties for violating their provisions, including those related to confidentiality.

Consent Is Generally Not Required for Newborn Screening, but Many States Allow Religious Exemptions

All states require newborn screening, and state newborn screening statutes usually do not require consent for screening. Only Wyoming's newborn screening statute expressly requires that persons responsible for collecting the blood specimen obtain consent prior to screening. In addition, of the three states with only regulations requiring newborn screening,[38] Maryland's regulations on newborn screening require consent for screening.[39]

While all states require newborn screening, most newborn screening statutes or regulations provide exemptions in certain situations. In 33 states, newborn screening statutes or regulations provide an exemption from screening if it is contrary to parents' religious beliefs or practices. Thirteen additional states provide an exemption for any reason. (See table 5.)

Table 5: Basis on Which Newborn Screening Exemption Is Granted, by State

	Basis for exemption		
	Religious objection	Any objection	No exemption
Alabama	X		
Alaska		X	
Arizona			X
Arkansas	X		
California	X		
Colorado		X	
Connecticut	X		
Delaware	X		
District of Columbia		X	
Florida		X	
Georgia	X		
Hawaii	X		
Idaho	X		
Illinois	X		
Indiana	X		
Iowa		X	
Kansas	X		
Kentucky	X		

[38]These states do not have newborn screening statutes.

[39]Both Wyoming's newborn screening statute and Maryland's newborn screening regulation expressly require informed consent; however, neither state's newborn screening statute or regulation defines this term.

	Basis for exemption		
	Religious objection	Any objection	No exemption
Louisiana		X	
Maine	X		
Maryland		X	
Massachusetts	X		
Michigan	X		
Minnesota	X		
Mississippi	X		
Missouri	X		
Montana			X
Nebraska			X
Nevada		X	
New Hampshire		X	
New Jersey	X		
New Mexico		X	
New York	X		
North Carolina		X	
North Dakota	X		
Ohio	X		
Oklahoma	X		
Oregon	X		
Pennsylvania	X		
Rhode Island	X		
South Carolina	X		
South Dakota			X
Tennessee	X		
Texas	X		
Utah	X		
Vermont		X	
Virginia	X		
Washington	X		
West Virginia			X
Wisconsin[a]	X		
Wyoming		X	

Sources: State newborn screening statutes and newborn screening regulations.

Note: GAO analysis of state newborn screening statutes and newborn screening regulations.

[a]Wisconsin's screening statute also authorizes a urine test program to test infants for causes of congenital disorders, but provides that no person may be required to participate in that program.

Most States Have Privacy Laws or Regulations That Protect Newborn Screening Information to Some Extent

In over half the states, newborn screening statutes and regulations have provisions that indicate that information collected from newborn screening is confidential.[40, 41] However, they permit information to be released without authorization from the child's legal representative in some circumstances. The most common provision for release of screening information is for use in statistical analysis or research, generally with a requirement that the identity of the subject is not revealed and/or that the researchers comply with applicable state and federal laws for the protection of humans in research activities. Some state screening statutes have additional provisions that allow screening information to be released. Wisconsin's screening statute, for example, allows the information to be released for use by health care facilities staff and accreditation organizations for audit, evaluation, and accreditation activities; and for billing, collection, or payment of claims. A few states have more restrictive provisions. South Carolina's screening statute, for example, limits disclosure of the information obtained from screening to the physician, the parents of the child, and the child when he or she reaches age 18.

State statutes that govern the collection, use, or disclosure of genetic information may also apply to genetic information obtained from newborn screening. Twenty-five states have laws that prohibit disclosure of genetic information without the consent of the individual; in 23 of these states, the statutes have exceptions that permit disclosure without consent.[42] (See table 6.) For example, 14 states' genetic privacy laws permit disclosure of genetic information without consent for the purpose of research, provided that individuals' identities are not revealed and/or the research complies with applicable state and federal laws for the protection of humans in research activities.

[40]We found no limitation on the ability of laboratories or state agencies to inform health care providers attending newborns with abnormal screening results. On the contrary, many statutes and regulations require laboratories and state agencies to inform providers of abnormal screening results.

[41]As defined in federal regulations implementing the Health Insurance Portability and Accountability Act of 1996, the term health information would also include newborn screening information.

[42]This analysis is based on National Conference of State Legislatures' information indicating that 29 states have laws that govern the privacy of genetic information. In 4 of these states, the statutes relate only to collection and/or use of genetic information.

Table 6: Exceptions to Confidentiality Requirements in States' Genetic Privacy Laws

State with genetic privacy law	Exception				
	Research[a]	Disclosure to health care provider	Peer review or quality assurance activity	Establishing paternity	In connection with law enforcement or legal proceedings
Arizona	X	X	X		
Arkansas	X				
Colorado	X			X	
Delaware				X	X
Florida				X	X
Georgia	X				X
Illinois		X	X	X	X
Louisiana	X			X	X
Maryland	X	X			
Massachusetts	X				X
Missouri	X				X
Nevada		X		X	X
New Hampshire		X		X	X
New Jersey				X	X
New Mexico	X	X		X	X
New York					X
Oregon	X			X	X
Rhode Island	X				
South Carolina				X	X
Texas	X			X	X
Utah					X
Vermont	X			X	X
Washington	X	X	X		

Sources: State statutes.

Notes: GAO analysis of state statutes. States' genetic privacy laws may also apply to genetic information obtained from newborn screening.

[a]Information may be disclosed for research, subject to conditions concerning the release of individuals' identities and/or compliance with state and federal laws for the protection of humans in research activities.

Most state newborn screening statutes and genetic privacy laws do not include penalties for lack of compliance. According to the National Conference of State Legislatures, 17 states have laws that provide specific penalties for violating genetic privacy laws. In 6 of these states, violations of genetic privacy statutes are punishable by fine and/or imprisonment. In addition, the statutes authorize civil lawsuits to obtain damages and, in most instances, court costs and attorneys' fees. In 10 of these states, the

statutes provide for civil liability only. In 1 state, violation is punishable only as a crime.

Agency Comments

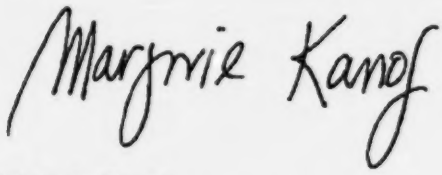
We provided a draft of this report to HHS for comment. Overall, HHS said that the report presents a thorough summary of state newborn screening programs' current practices. (HHS's comments are reprinted in app. VI.) HHS said that the report needed to reflect that newborn screening is a system that, in addition to testing, includes follow-up, diagnosis, disease management and treatment, evaluation, and education. However, the draft report did identify the various components of the newborn screening system. HHS said that there is a need to more comprehensively address components of the system beyond testing. For example, HHS commented that there is a need for a coordinated effort in states to train and educate health professionals and state newborn screening program directors in the use of newer technologies. In addition, it stated that there is a need to provide information to families and parents about the screening their state provides and the screening options available to them outside of their state's program. HHS said that it anticipated that the report would, among other things, include recommendations to improve state newborn screening programs. As we noted in the draft report, HRSA has initiated a process to develop recommendations for state newborn screening programs. The scope of our review focused on providing the Congress with descriptive information about state programs.

HHS supported the development of benchmarks to help states evaluate the quality of the various components of the newborn screening system. It added that one of the most effective ways the federal government can support state newborn screening programs is by strengthening the scientific basis for newborn screening through funding of systematic evaluation of outcomes and the quality of all components of the newborn screening system.

In its comments, HHS provided information on its efforts related to newborn screening. For example, HHS described demonstration projects it funded to examine the use of new technology and initiatives to improve family and provider education. In addition, HHS indicated that all of its programs address the recommendations of the AAP newborn screening task force and encourage the integration of various newborn screening and genetics services into systems of care. HHS provided technical comments. We incorporated the technical comments and other information HHS provided on its programs where appropriate.

As arranged with your offices, unless you publicly announce its contents earlier, we will not distribute this report until 30 days after its issue date. We will then send copies of this report to the Secretary of Health and Human Services, the Administrators of the Health Resources and Services Administration and the Centers for Medicare & Medicaid Services, the Directors of the Centers for Disease Control and Prevention and the National Institutes of Health, appropriate congressional committees, and others who are interested. We will also make copies available to others upon request. In addition, the report will be available at no charge on the GAO Web site at http://www.gao.gov.

If you or your staff have any questions, please contact me at (202) 512-7119. An additional contact and the names of other staff members who made contributions to this report are listed in appendix VII.



Marjorie Kanof
Director, Health Care—Clinical
and Military Health Care Issues

Appendix I: Scope and Methodology

To do our work, we surveyed the health officers in all the states during October and November 2002 about their newborn screening programs.[1] We asked each state health officer to work with laboratory and program administration/follow-up staff in responding to the questions. The survey asked for information on the process for selecting disorders to include in newborn screening programs; laboratory and follow-up activities; parent and provider education efforts; expenditures and funding sources; efforts to evaluate the quality of laboratory testing and program administration/follow-up; and states' retention and sharing of screening results. The survey focused only on screening for metabolic and genetic disorders. We did not ask for information on disease management and treatment services provided by state newborn screening programs, and the survey did not collect information on newborn screening for hearing and infectious diseases.

We pretested the survey in person with laboratory and program administration/follow-up staff from the Virginia and Delaware newborn screening programs. In addition, the survey instrument was reviewed by staff at the Department of Health and Human Services' (HHS) Centers for Disease Control and Prevention (CDC), National Center for Environmental Health, Newborn Screening Branch, and the National Newborn Screening and Genetics Resource Center, a project funded by HHS's Health Resources and Services Administration (HRSA). We refined the questionnaire in response to their comments. We received responses from all the states. After reviewing the completed questionnaires and checking the data for consistency, we contacted certain states to clarify responses and edited survey responses as appropriate. In addition, we followed up with four states to obtain more detailed information on their processes for selecting disorders, evaluations of parent and provider education, evaluations of the quality of laboratory testing and program administration/follow-up, and mechanisms for identifying abnormal cases misclassified as normal.

To identify which genetic and metabolic disorders are included in states' newborn screening programs, we reviewed the Resource Center's U.S. National Screening Status Reports. These reports provide information on the disorders for which states require screening and the disorders for which screening is provided to selected populations, through pilot programs, or by request.

[1]"States" refers to the 50 states and the District of Columbia.

To report on efforts by HHS and states to monitor and evaluate the quality of state newborn screening programs, we reviewed annual summary reports, proficiency testing results, and other documents from the Newborn Screening Quality Assurance Program (NSQAP), which CDC operates with the Association of Public Health Laboratories, and interviewed CDC staff on states' participation. We also reviewed report findings from the seven technical reviews of state newborn screening programs that HRSA, CDC, and the Resource Center conducted from 1999 to 2001. We interviewed Resource Center staff about the content and findings of these reviews and interviewed officials in five states about actions taken in response to the review staff's findings and recommendations.

To determine how state laws address consent and privacy issues related to newborn screening, we analyzed state statutes that provide for newborn screening for genetic and metabolic disorders and state statutes that relate to privacy of genetic information generally. We also reviewed state newborn screening regulations as appropriate. The information on states that require consent for newborn screening is based on our analysis of state newborn screening and genetic privacy statutes and the newborn screening regulations in states that do not have newborn screening statutes. The information on exemptions from screening is based on our review of state newborn screening statutes and newborn screening regulations. Information on privacy is based on our analysis of confidentiality provisions in state newborn screening statutes and, for those states that do not have confidentiality provisions in their newborn screening statutes, on confidentiality provisions in newborn screening regulations. We also analyzed confidentiality provisions in state genetic privacy statutes.

To identify the newborn screening statutes and regulations that were within the scope of our review, we relied on research provided by the National Conference of State Legislatures (NCSL) in fall 2002 and analyzed only those newborn screening statutes and regulations identified through that research. With regard to genetic privacy statutes, we analyzed only those statutes identified by NCSL in an April 2002 report identifying state genetic privacy laws.[2] We contacted state officials as appropriate to obtain assistance in locating and interpreting statutory authorities. We also relied

[2]National Conference of State Legislatures, *Genetics Policy Report, Privacy* (Washington, D.C.: April 2002).

on NCSL's determination of the number of states that provide penalties for the violation of those statutes.

Newborn screening programs are governed by a variety of legal authorities. We did not research or analyze any case law interpreting state newborn screening statutes and regulations or genetic privacy statutes, and we did not research or analyze any written interpretive guidance issued by states.

We also reviewed relevant literature and obtained information from individual experts, newborn screening laboratory and maternal and child health staff in several states, and representatives of organizations interested in newborn screening, including the American Academy of Pediatrics, American College of Medical Genetics, American College of Obstetricians and Gynecologists, American Medical Association, Association of Maternal and Child Health Programs, Association of Public Health Laboratories, Association of State and Territorial Health Officials, and the March of Dimes.

We conducted our work from June 2002 through March 2003 in accordance with generally accepted government auditing standards.

Appendix II: Number of Disorders Included in State Newborn Screening Programs, December 2002

	Number of disorders		Number of disorders for which screening is conducted using tandem mass spectrometry (MS/MS)[a,b]	
	Screening required for all newborns	Screening conducted for selected populations, as pilot program, or by request	Screening required for all newborns	Screening conducted for selected populations, as pilot program, or by request
Alabama	5	0	0	0
Alaska	6	1	0	0
Arizona	8	0	0	0
Arkansas	4	0	0	0
California	4	28	0	28
Colorado	7	0	0	0
Connecticut	8	1	0	0
Delaware	5	0	0	0
District of Columbia	7	0	0	0
Florida	5	0	0	0
Georgia	8	0	0	0
Hawaii	7	28	0	28
Idaho	5	27	0	26
Illinois	27	0	19	0
Indiana	9	0	1	0
Iowa	6	30	1	27
Kansas	4	0	0	0
Kentucky	4	0	0	0
Louisiana	5	0	0	0
Maine	9	18	1	18
Maryland	9	0	0	0
Massachusetts	10	20	1	19
Michigan	7	0	0	0
Minnesota	5	21	0	19
Mississippi	5	0	0	0
Missouri	5	0	0	0
Montana	3	18	0	14
Nebraska	5	28	0	26
Nevada	6	0	0	0
New Hampshire	6	1	0	0
New Jersey	14	0	6	0
New Mexico	6	0	0	0
New York	10	0	1	0
North Carolina	32	0	25	0
North Dakota	4	2	0	1
Ohio	12	15	6	15
Oklahoma	4	0	0	0
Oregon	33	0	26	0

	Number of disorders		Number of disorders for which screening is conducted using tandem mass spectrometry (MS/MS)[a,b]	
	Screening required for all newborns	Screening conducted for selected populations, as pilot program, or by request	Screening required for all newborns	Screening conducted for selected populations, as pilot program, or by request
Pennsylvania	6	0	0	0
Rhode Island	9	0	1	0
South Carolina	6	0	1	0
South Dakota	3	29	0	26
Tennessee	5	0	0	0
Texas	5	0	0	0
Utah	4	0	0	0
Vermont	7	0	0	0
Virginia	8	0	0	0
Washington	4	0	0	0
West Virginia	3	1	0	0
Wisconsin	21	5	14	3
Wyoming	6	0	0	0

Source: National Newborn Screening and Genetics Resource Center websites: http://genes-r-us.uthsca.edu/resources/newborn/screenstatus.htm, downloaded on January 9, 2003, and http://genes-r-us.uthsca.edu/resources/newborn/msmstests.htm, downloaded on January 8, 2003.

[a]States may use their own laboratory to conduct MS/MS screening or contract with other laboratories.

[b]Numbers exclude MS/MS screening for phenylketonuria, maple syrup urine disease, and homocystinuria.

Appendix III: Information on Disorders Most Commonly Included in State Newborn Screening Programs

Disorder	National incidence[a]	Description	Potential outcomes	Treatment
Phenylketonuria	1 in 13,947[b]	Deficiency of an enzyme needed to break down the amino acid phenylalanine	Mental retardation, seizures	Low-phenylalanine diet
Congenital hypothyroidism	1 in 3,044[c]	Inability to produce adequate amount of thyroid hormone	Mental retardation, stunted growth	Thyroid hormone
Galactosemia	1 in 53,261[d]	Deficiency of an enzyme needed to break down the milk sugar galactose	Brain damage, liver damage, cataracts, death	Galactose-free diet
Sickle cell diseases	1 in 3,721/ 1 in 7,386[e]	Inherited blood disorder causing hemoglobin abnormalities	Organ damage, delayed growth, stroke	Penicillin, vaccinations
Congenital adrenal hyperplasia	1 in 18,987	Deficiency of an adrenal enzyme needed to produce cortisol and aldosterone	Death due to salt loss, reproductive and growth difficulties	Hormone replacement and salt replacement
Biotinidase deficiency	1 in 61,319	Deficiency of the enzyme biotinidase, needed to recycle the vitamin biotin	Mental retardation, developmental delay, seizures, hearing loss	Biotin supplements
Maple syrup urine disease	1 in 230,028	Deficiency of the enzyme needed to metabolize leucine, iscleucine, and valine	Mental retardation, seizures, coma, death	Dietary management and supplements
Homocystinuria	1 in 343,650	Deficiency of the enzyme needed to metabolize the amino acid homocysteine	Mental retardation, eye problems, skeletal abnormalities, stroke	Dietary management and vitamin supplements

Sources: National Newborn Screening and Genetics Resource Center and newborn screening literature.

[a]Preliminary data on disorder incidence presented by the National Newborn Screening and Genetics Resource Center at the 2002 Newborn Screening and Genetic Testing Symposium. Incidence rates are based on data from 1990 to 1999.

[b]Incidence rate is for clinically significant hyperphenylalaninemia, which includes classical phenylketonuria and clinically significant phenylketonuria variant.

[c]Incidence rate is for primary congenital hypothyroidism and does not include other forms of hypothyroidism.

[d]Incidence rate is for classical galactosemia and does not include other forms of galactosemia.

[e]Sickle cell anemia has an incidence of 1 in 3,721, while Hemoglobin sickle C disease has an incidence of 1 in 7,386.

Appendix IV: Selected Disorders States Screen for Using MS/MS and Number of States That Screen for Each, December 2002

Disorder	Number of states[a]	
	Screening required for all newborns	Screening conducted for selected populations, as pilot program, or by request
Fatty acid oxidation defects		
Carnitine palmitoyl transferase deficiency type I (CPT-1)	2	4
Carnitine palmitoyl transferase deficiency type II (CPT-2)	4	11
Carnitine/acylcarnitine translocase deficiency (CAT)	3	8
Long-chain hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)	4	11
Multiple acyl-CoA dehydrogenase deficiency (GA-II)	4	11
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)	5	11
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	13	8
Trifunctional protein deficiency	3	8
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	4	11
Long-chain acyl-CoA dehydrogenase deficiency (LCAD)	4	6
2,4 dienoyl-CoA reductase deficiency	2	2
Organic acidemias		
Glutaric aciduria type I (GA-1)	4	11
3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)	4	11
Isobutyryl-CoA dehydrogenase deficiency	1	6
Isovaleric acidemia (IVA)	5	10
Malonic aciduria	0	5
3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)	4	11
Methylmalonic acidemia (MMA)	5	10
Mitochondrial acetoacetyl-CoA thiolase deficiency (3-ketothiolase)	3	10
Propionic acidemia (PA)	5	10
2-methylbutyrl-CoA dehydrogenase deficiency	2	6
Multiple CoA carboxylase deficiency	1	4
Other amino acidemias		
Argininemia	2	10
Argininosuccinate lyase deficiency (ASA)	5	10
Citrullinemia	5	10
Hyperammonemia, hyperornithinemia, homocitrullinuria (HHH)	2	8
Nonketotic hyperglycinemia	1	6
5-oxoprolinuria	1	4
Tyrosinemia type I	3	10
Tyrosinemia type II	2	7

Source: National Newborn Screening and Genetics Resource Center website, http://genes-r-us.uthsca.edu/resources/newborn/msmstests.htm, downloaded January 14, 2003.

[a]"States" refers to the 50 states and the District of Columbia.

Appendix V: State Newborn Screening Program Fees and Expenditures Per Infant Screened

	Newborn screening fee[a]	Expenditures per infant screened[b]
Alabama	$34.00	$32.11[c]
Alaska	24.00	28.78
Arizona	20.00/20.00[d]	25.99[e]
Arkansas	14.83	17.95
California	60.00	50.85
Colorado	43.47	30.63
Connecticut	28.00	39.20
Delaware	40.69	61.28
District of Columbia	No fee	25.96
Florida	20.00	[f]
Georgia	No fee	[f]
Hawaii	27.00	26.65
Idaho	18.00	16.11
Illinois	32.00	31.00
Indiana	39.50	28.16[c]
Iowa	46.00	32.73
Kansas	No fee	17.37
Kentucky	14.50	[f]
Louisiana	18.00	25.62
Maine	33.00	34.37
Maryland	30.00	30.90[c]
Massachusetts	49.55	50.12
Michigan	42.61	25.69[c]
Minnesota	21.00	[f]
Mississippi	25.00	25.00
Missouri	25.00	26.02
Montana	36.92	48.35
Nebraska	50.00/54.60[g]	44.01
Nevada	30.00	22.96
New Hampshire	18.00	22.24
New Jersey	34.00	38.27[c]
New Mexico	32.00	31.59
New York	No fee	39.92
North Carolina	10.00	14.75
North Dakota	18.00	20.81
Ohio	33.75	21.77[c]
Oklahoma	10.50	23.43
Oregon	27.00	25.05
Pennsylvania	No fee	19.91
Rhode Island	59.00	38.52
South Carolina	21.00	38.28

	Newborn screening fee[a]	Expenditures per infant screened[b]
South Dakota	No fee	
Tennessee	17.50	19.34
Texas	19.50	19.74
Utah	31.00	19.62
Vermont	27.00	27.60
Virginia	27.00	30.89
Washington	40.40	39.31
West Virginia	No fee	15.98
Wisconsin	59.50	33.35
Wyoming	No fee	16.23

Source: GAO Survey of State Newborn Screening Programs for Genetic and Metabolic Disorders, October 21, 2002.

[a]We asked states to report their current fee. States responded to the survey in October and November 2002.

[b]State fiscal year 2001.

[c]State's expenditures per infant screened may not reflect a typical year because the state reported that its expenditures for state fiscal year 2001 included a significant, nonrecurring expenditure.

[d]State charges two fees, one at initial screening and another at the second screening.

[e]Expenditures include disease management and treatment services.

[f]Expenditure per infant screened not calculated because state did not report number of infants screened.

[g]Fee varies depending on laboratory conducting the screening.

[h]Expenditure information not available for state fiscal year 2001.

Appendix VI: Comments from the Department of Health and Human Services

DEPARTMENT OF HEALTH & HUMAN SERVICES

Office of Inspector General

Washington, D.C. 20201

MAR 6 2003

Ms. Marjorie E. Kanof
Director, Health Care -- Clinical
and Military Health Care Issues
United States General
Accounting Office
Washington, D.C. 20548

Dear Ms. Kanof:

Enclosed are the department's comments on your draft report entitled, "Newborn Screening: Characteristics of State Programs." The comments represent the tentative position of the department and are subject to reevaluation when the final version of this report is received.

The department also provided several technical comments directly to your staff.

The department appreciates the opportunity to comment on this draft report before its publication.

Sincerely,

Janet Rehnquist

Janet Rehnquist
Inspector General

Enclosure

The Office of Inspector General (OIG) is transmitting the department's response to this draft report in our capacity as the department's designated focal point and coordinator for General Accounting Office reports. The OIG has not conducted an independent assessment of these comments and therefore expresses no opinion on them.

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Comments of the Department of Health and Human Services on the General Accounting Office's Draft Report, "Newborn Screening: Characteristics of State Programs" (GAO-03-449)

The Department of Health and Human Services (HHS) appreciates the opportunity to comment on the GAO's draft report, "Newborn Screening: Characteristics of State Programs." Overall, the report presents a thorough summary of the current practices and the successes of and challenges to newborn screening programs. A summary of general comments from the Office of the Assistant Secretary for Planning and Evaluation, the Health Resources and Services Administration (HRSA), the Centers for Disease Control and Prevention (CDC), and the National Institutes of Health (NIH) are presented below.

General Comments

Newborn screening is more than a screening test; this conceptual framework needs to be reflected in the report. The screening test is only part of a newborn screening system that includes follow-up, diagnosis, management, treatment, evaluation, and education. In the coming years, there will be a need to more comprehensively address these additional components of the system. For example, there is a need for concerted, coordinated effort at the state levels to train and educate health professionals and state newborn screening program directors in the use of newer technologies such as tandem mass spectrometry. A similar effort needs to be made with families and parents. They need to understand what newborn screening is, how it is done, who performs it, and where. They also need information about the medical conditions their state mandates and offers in its screening program, and the options available for screening for conditions in addition to those for which the state screens.

The HHS anticipated a report that would review the functioning of state newborn screening programs, identify gaps, and make recommendations for improvements to ensure the health of our nation's children. Although the GAO report does point to the lack of uniformity between states, HHS would hope that the report's recommendations also address identified needs listed above. In addition, HHS would like to see benchmarks which states could use to evaluate the various components of the newborn screening system (and although beyond the scope of the report, including treatment and long-term follow-up). Both the National Committee for Clinical Laboratory Standards and the HRSA-funded Council of Regional Genetic Services Networks (CORN) have published national standards for the various components of newborn screening, including standards for specimen collection for newborn screening programs. The HHS would like to see additional work on the application of these standards. The following is an example of an area that might have been addressed in greater detail:

- Turnaround Time – Little is known regarding the time to the receipt of repeat specimens in cases where the initial specimen was either not collected, was improperly collected, or resulted in an abnormal initial screen.

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The HHS believes strengthening the scientific basis for newborn screening through funding of systematic evaluation of outcomes and the quality of all components of the newborn screening system is one of the most effective ways the federal government can support state newborn screening programs.

HHS Support of State Newborn Screening Programs

The HHS has supported the development of newborn screening programs since the 1960's. Research at both NIH, specifically that of the National Institute of Child Health and Human Development and the HRSA's Maternal and Child Health Bureau (MCHB) have provided the groundwork for newborn screening. The National Center for Environmental Health's Newborn Screening Quality Assurance Program at CDC has served to facilitate quality assurance of the state laboratories. The CDC's National Center on Birth Defects and Developmental Disabilities (NCBDDD) provides expertise in the areas of epidemiologic surveillance and evaluation. In collaboration with research, laboratory quality assurance and surveillance activities supported by NIH and CDC, the discretionary grants portion of HRSA's Maternal and Child Health Block Grant Program, which is funded under Title V of the Social Security Act as a Special Project of Regional and National Significance (SPRANS), has borne primary responsibility for funding federal newborn and other genetic screening, counseling, and information projects.

In 1981 newborn screening programs became a SPRANS funding category. For a 3-year period during the mid-1980s, MCHB state grant allotments were also supplemented to encourage statewide newborn screening for sickle cell disease, which led to the rapid spread of these programs. Federal funding was welcomed by the states, whose health department budgets rarely covered such activities. By 1990, all 50 states, the District of Columbia, Puerto Rico, and the Virgin Islands had begun to develop statewide capacity for testing, counseling, education, and referral for sickle cell and other genetic disorders. By the late 1990's all states had their own newborn screening laws. As a result of the laws mandating phenylketonuria (PKU) testing and the establishment of health department newborn heelstick screening units, state newborn heelstick screening programs evolved with the goal of providing safe screening tests and appropriate follow-up to every newborn. In 1998, HRSA requested the American Academy of Pediatrics to convene a Newborn Screening Task Force (co-sponsored by NIH, CDC, the Agency for Healthcare Research and Quality [AHRQ], and national public health organizations), which provided recommendations on:

- The use of new and evolving technologies such as mass spectrometry and DNA-based technologies in newborn screening programs. These technologies enable screening for many conditions for which there is no effective treatment.
- Uniformity of standards across states to assure access to screening tests and procedures that meet national standards and guidelines.
- Strategies for family/public information about newborn screening.

All of HHS' programs are addressing the Task Force recommendations and have been developed to encourage the integration of various types of federal, state, and community

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funded newborn screening and genetics services into systems of care that are responsive to the individual needs of the people being served.

Relevant to newborn screening, most HHS programs (HRSA's MCHB and CDC's Newborn Screening Quality Assurance Program [NSQAP] and NCBDDD) fund state public health agencies and have included:

Technical Assistance to States

In fiscal year (FY) 2001, MCHB began funding a National Newborn Screening and Genetics Resource Center (Center). This Center is funded under a cooperative agreement with the University of Texas Health Science Center, San Antonio. The Center provides technical assistance to the states around newborn screening and genetics issues. The Center also provides a forum for interaction between consumers, health care professionals, researchers, organizations, and policy makers involved in refining and developing public health newborn screening and genetics programs. In addition, through its website, http://genes-r-us.uthscsa.edu/, it serves as a national resource for information and education on newborn screening and public health genetics.

All state newborn screening laboratories voluntarily participate in CDC's NSQAP for proficiency testing and quality assurance. The NCBDDD is currently devoting staff resources to studies of the effectiveness and cost-effectiveness of newborn screening and is working in collaboration with state governments on these studies. The CDC staff has published studies assessing the benefits of newborn screening for a range of disorders, including metabolic disorders, sickle cell disease, and cystic fibrosis. The CDC is also currently funding four states to conduct long-term follow up of children identified through newborn screening programs.

Guidelines for Uniformity

In FY 2001, MCHB contracted with the American College of Medical Genetics (ACMG) to convene a group of experts to standardize outcomes and guidelines for state newborn screening programs, and to define responsibilities for collecting and evaluating outcome data. The MCHB expects this contract to produce a recommendation for a uniform panel of conditions for states to adopt in their newborn screening programs. Representatives from HRSA, NIH, CDC and AHRQ, as organizational liaisons, are participating in this process. In addition, HHS has issued a charter for an Advisory Committee on Heritable Disorders and Genetic Diseases in Infants and Children, which was authorized by the Children's Health Act of 2000 to provide advice to the Secretary on newborn screening issues. The committee will be informed by the report of the ACMG expert group, but substantial further work may be needed prior to the issuance of recommendations to states. The HHS supports the development of federal screening recommendations that will be preceded by rigorous systematic reviews of scientific evidence conducted under open peer review.

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Use of New Technology

In FY 2001, MCHB funded demonstration projects to identify strategies and develop materials for examining the clinical validity and utility of new and emerging technologies within newborn screening programs.

In 2003, HRSA and the National Newborn Screening and Genetics Resource Center have conducted six sessions to train state newborn screening program laboratory scientists and program coordinators in the use of tandem mass spectrometry. These sessions are co-sponsored by CDC and the Association of Public Health Laboratory Directors.

Family Education

In FY 2001 and 2002, MCHB funded cooperative agreements with the March of Dimes and the Genetic Alliance to educate families about newborn screening and genetics.

Coordination

In FY 1998, MCHB began a pilot initiative to stimulate the integration of newborn screening programs and their information systems among the hospitals, the state laboratories and diagnostic centers, the infant's medical home, and the subspecialists who give the infant further diagnosis and treatment. These pilot projects have enhanced states' capacity to monitor access to care and outcomes; and to better integrate state newborn screening programs with the state Title V system of services for children with special health care needs, thereby better coordinating the delivery of early treatment and intervention.

Partnerships among Health Care Professionals

Since FY 1999, MCHB has funded the American Academy of Pediatrics to educate its membership about newborn screening. The Academy maintains a member website dedicated to screening.

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Appendix VII: GAO Contact and Staff Acknowledgments

GAO Contact

Helene F. Toiv, (202) 512-7162

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In addition to the person named above, key contributors to this report were Janina Austin, Emily Gamble Gardiner, Ann Tynan, Ariel Hill, Kevin Milne, Cindy Moon, and Susan Lawes.

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